Congenital Infiltrating Lipomatosis of the Cheek in an Adolescent: A Case Report and Literature Review

Abstract:

Background: Congenital infiltrating lipomatosis of the face (CILF) is a rare, histologically benign but locally infiltrative disorder characterised by a diffuse proliferation of mature, non-encapsulated adipose tissue that invades the facial soft tissues and, in some patients, the underlying bone and teeth.

Methods: We report a case managed at Oued Eddahab Military Hospital, Agadir, Morocco. Clinical, radiological, operative and histopathological data were reviewed and compared with the literature.

Results: A 15-year-old boy presented with a slowly progressive swelling of the left cheek, present since childhood and responsible for facial asymmetry. Magnetic resonance imaging (MRI) showed diffuse fatty infiltration of the left buccal soft tissues involving the buccal fat pad and extending into the masticator space, without significant enhancement. Conservative partial excision was performed through an extended pretragal approach combined with a nasolabial fold incision. Histopathology showed mature adipose tissue without atypia or malignancy. The postoperative course was uneventful, with no facial nerve deficit and no salivary fistula.

Conclusion: CILF should be considered in any soft, ill-defined, unilateral cheek swelling that has progressed since childhood. MRI is essential for mapping the lesion, and surgery should be conservative and individualised, aiming at aesthetic and functional improvement while preserving the facial nerve and the parotid duct. Long-term follow-up is mandatory because of the risk of recurrence.

Keywords: congenital infiltrating lipomatosis; cheek; facial asymmetry; magnetic resonance imaging; PIK3CA; case report.

Introduction

Congenital infiltrating lipomatosis of the face (CILF) is a rare entity first described by Slavin et al. in 1983 [1]. It consists of a diffuse proliferation of mature, non-encapsulated adipocytes that infiltrate the adjacent muscles and soft tissues, without evidence of malignancy [1,2]. Unlike a conventional lipoma, it has ill-defined margins, tends to extend progressively and is rarely amenable to complete excision. The cheek is one of the most frequently involved sites, and the disease usually presents as a unilateral facial asymmetry noticed at birth or in early childhood [2,3]. Depending on the extent of the lesion, dental, osseous, mucosal or parotid abnormalities may be associated [4–6]. The diagnosis relies on the correlation of clinical, radiological and histopathological findings, magnetic resonance imaging (MRI) being the reference investigation for characterising the fatty component and its extension [3].

We report a case of CILF of the left cheek in an adolescent and discuss its diagnostic, therapeutic and prognostic aspects in the light of the current literature.

Materials and Methods

This work is a single-patient case report combined with a narrative review of the literature, prepared in accordance with the CARE (CAse REport) guidelines. The patient was managed in the Department of Oral and Maxillofacial Surgery and Stomatology of Oued Eddahab Military Hospital, Agadir, Morocco. Data were retrieved from the medical record and included the history of the disease, extraoral and intraoral examination, MRI findings, the operative report, the postoperative course and the histopathological report.

The clinical variables recorded were age, sex, medical history, duration of progression, characteristics and morphological impact of the swelling, condition of the oral mucosa, patency of the parotid (Stensen's) duct orifice, and examination of the parotid gland, cranial nerves and cervical lymph nodes. MRI was analysed for signal intensity, suppression on fat-saturated sequences, enhancement after gadolinium administration, and the relationship of the lesion with the buccal fat pad and the masticator space. The literature review covered clinical series, imaging features, dentoskeletal abnormalities, genetic data and therapeutic options in CILF. Written informed consent for publication, including clinical images, was obtained from the patient's legal representative, and all images were anonymised.

Results: Case Presentation

Patient information

A 15-year-old boy with no relevant medical history presented with a swelling of the left cheek that had been progressing slowly since childhood. There had been no inflammatory episode, spontaneous pain, bleeding or sign of infection. He reported no sensory disturbance, no feeding difficulty and no episode of facial palsy. The main complaint was facial asymmetry, which had become more conspicuous with growth.

Clinical findings

Extraoral examination revealed a soft, ill-defined, painless, non-inflammatory, non-pulsatile and non-compressible swelling of the lower left cheek, measuring approximately 3 × 6.5 cm, with downward displacement of the left labial commissure (Figure 1). The overlying skin was normal, with no erythema, local warmth or visible superficial venous network. Intraoral examination showed bulging of the left buccal mucosa without ulceration or mucosal infiltration; the parotid duct orifice was patent and non-inflamed. The parotid gland was not enlarged. Examination of the cranial nerves, including facial nerve function, was normal, and the cervical lymph node areas were free.

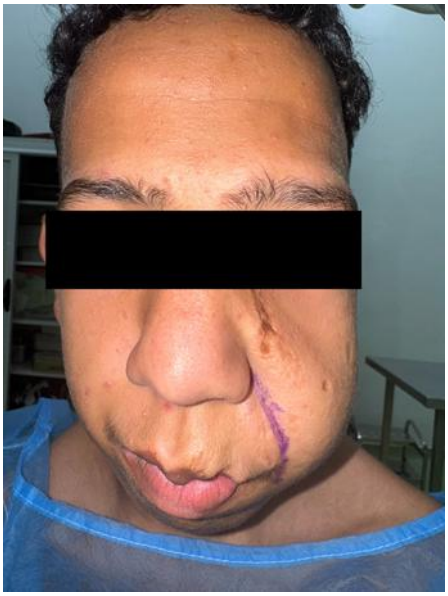

Figure 1. Preoperative frontal view showing infiltrative hypertrophy of the lower left cheek responsible for facial asymmetry and downward displacement of the left labial commissure; the planned nasolabial fold incision is marked on the skin. The periorbital region has been masked to preserve anonymity.

Diagnostic assessment

Facial MRI showed an ill-defined lesion with a fatty signal, hyperintense on T1- and T2-weighted images, with signal suppression on fat-saturated sequences and no significant enhancement after gadolinium injection. The lesion diffusely infiltrated the left buccal soft tissues, involved the buccal fat pad and extended into the masticator space (Figure 2). These features favoured infiltrating lipomatosis over a vascular or lymphatic malformation.

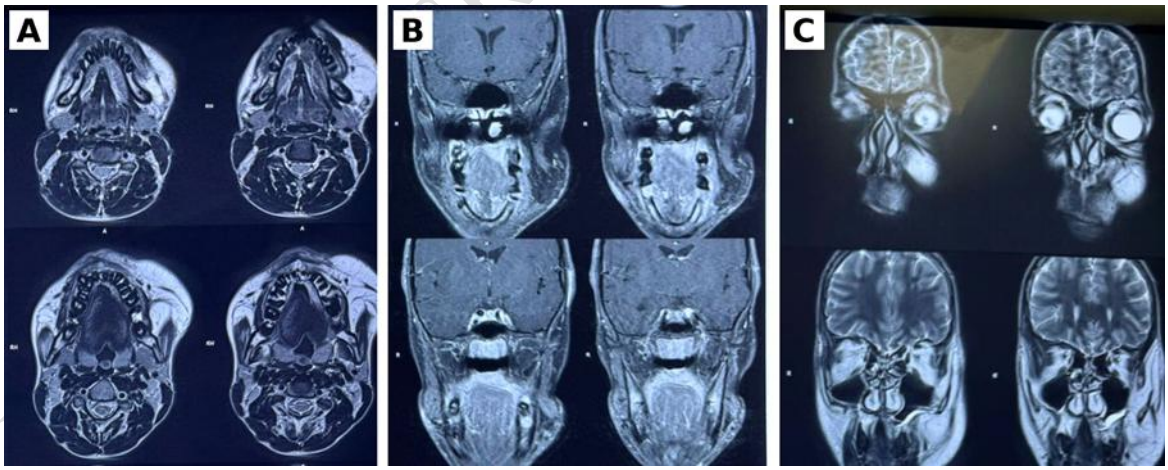

Figure 2. Facial MRI. (A) Axial slices showing diffuse fatty infiltration of the left buccal soft tissues. (B) Coronal fat-suppressed slices showing signal suppression of the lesion and its extension towards the deep spaces of the face. (C) Additional coronal sequences confirming the buccal location and the fatty nature of the lesion.

Therapeutic intervention

Surgery was performed under general anaesthesia through two approaches: a left extended pretragal (preauricular) incision, providing access to the posterior buccal region and the masseteric space, and a nasolabial fold incision to control the anterior component. After

dissection in the subcutaneous plane superficial to the superficial musculoaponeurotic system (SMAS), the lesion appeared as yellowish, diffuse, non-encapsulated adipose tissue infiltrating the buccal muscle planes without a cleavage plane (Figure 3). A partial, stepwise, piecemeal resection was carried out; complete deep excision was deliberately not attempted in order to preserve the branches of the facial nerve, the parotid duct and perioral mobility.

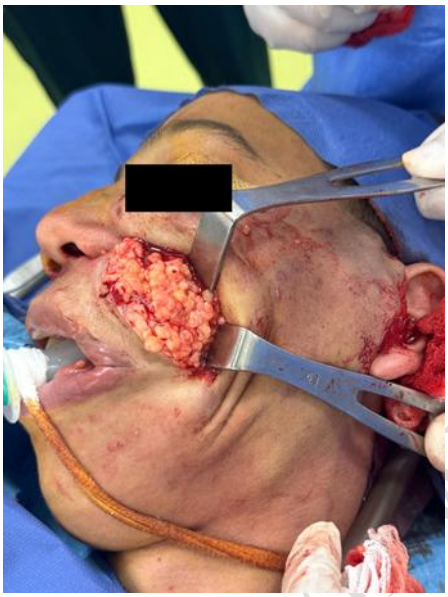

Figure 3. Intraoperative view showing diffuse infiltration of the left buccal region by yellowish, non-encapsulated adipose tissue.

Follow-up, outcome and histopathology

The immediate postoperative course was uneventful, without facial nerve deficit, salivary fistula or infection. Histopathological examination of the specimen showed a proliferation of mature adipocytes without lipoblasts or cytological atypia, consistent with a lipomatous lesion and with no evidence of malignancy. Given the diffuse, non-encapsulated, infiltrative and long-standing nature of the lesion, correlation of the clinical, radiological, operative and histological findings established the diagnosis of CILF of the left cheek. The timeline of the case is summarised in Table 1.

Table 1. Timeline of the patient's management (CARE guidelines).

Period	Clinical event
Birth – early childhood	Onset of a swelling of the lower left cheek.
Childhood – adolescence	Slow, painless progression without inflammatory or vascular signs; worsening facial asymmetry with growth.
Presentation (age 15 years)	Soft, ill-defined, painless, non-pulsatile mass (≈ 3 × 6.5 cm) with downward displacement of the left labial commissure.
Diagnostic work-up	MRI: diffuse fatty infiltration of the left buccal soft tissues, buccal fat pad and masticator space, without significant enhancement.
Surgery	Conservative partial piecemeal excision under general anaesthesia through extended pretragal and nasolabial fold approaches.
Early postoperative course	Uneventful: no facial nerve deficit, no salivary fistula, no infection.

Period	Clinical event
Histopathology	Mature adipose proliferation without lipoblasts or atypia; no evidence of malignancy.
Final diagnosis and follow-up	CILF of the left cheek; long-term follow-up for recurrence, aesthetic outcome and perioral function.

CILF: congenital infiltrating lipomatosis of the face; MRI: magnetic resonance imaging.

Discussion

CILF occupies a particular place among adipose lesions of the head and neck: it is histologically benign but locally infiltrative, which explains the difficulty of its treatment. Its classic histological description comprises mature, non-encapsulated adipocytes infiltrating muscles and soft tissues, without lipoblasts or cytological atypia, sometimes accompanied by nerve fibres, fibrous septa and increased vascularity [1,2]. A pathology report of "lipoma" is therefore insufficient on its own and must be interpreted together with the clinical, radiological and operative findings. In our patient, the tissue was mature and benign but diffuse, ill-defined and infiltrative, which fits the spectrum of CILF rather than an encapsulated lipoma.

The age at diagnosis and the natural history of our case are also characteristic. Most patients present with facial asymmetry at birth or in early childhood, followed by slow progression during growth [2,4]. Paucisymptomatic forms may be neglected for a long time and managed only in adolescence, when the aesthetic impact becomes more pronounced. In our patient, the swelling had been present since childhood and progressed slowly without pain, inflammatory signs or vascular manifestations. This chronic, silent course is consistent with published reports, in which facial asymmetry rather than functional impairment is the main complaint [4,7].

The cheek is a frequent site of the disease, but involvement may extend to the lip, tongue, parotid gland, masticatory muscles, soft palate, maxilla, mandible or zygoma [5,6,8]. In recent series, involvement is usually hemifacial and generally respects the midline; the most extensive forms may be associated with macroglossia, labial thickening, epidermal naevi, bone hypertrophy and dental abnormalities (Figure 4) [8,9]. Our case is distinguished by a predominantly buccal presentation without macroglossia, visible mucosal involvement or neurological abnormality. This localised form nevertheless remains typical because of the unilateral, progressive and infiltrative nature of the mass.

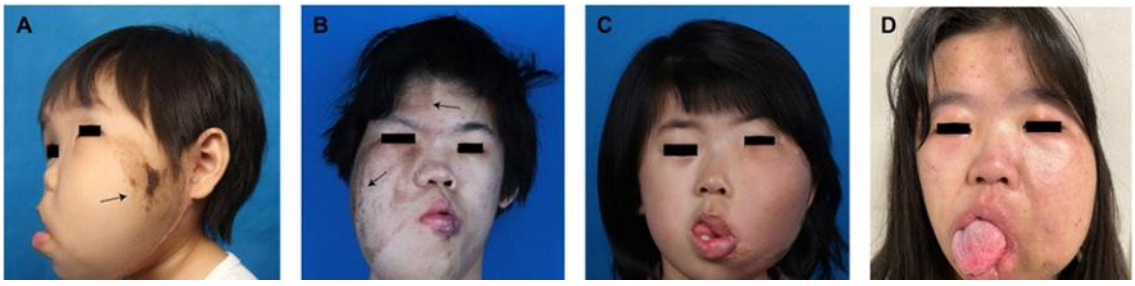

Figure 4. Clinical presentations of facial infiltrating lipomatosis illustrating phenotypic variability: epidermal naevi in the trigeminal dermatomes (A, B), labial thickening (C) and hemimacroglossia

with lower lip thickening (D). *Reproduced from Chen et al. [8], an open-access article distributed under the Creative Commons Attribution 4.0 International License (CC BY 4.0).*

Imaging is decisive for the diagnosis. Ultrasonography may be performed first, but it hardly distinguishes pathological fatty infiltration from normal buccal fat and is of limited value for assessing deep extension [3]. Computed tomography (CT) is useful for evaluating bony structures, dental abnormalities and skeletal hyperplasia. MRI is the most informative investigation for soft tissues: the lesion is hyperintense on T1- and T2-weighted images, with signal suppression on fat-saturated sequences and absent or faint enhancement after gadolinium administration [3]. These criteria help to exclude lymphatic or venous malformations, which may mimic a cheek swelling in a child. The MRI of our patient showed exactly this profile, with extension to the buccal fat pad and the masticator space.

Dental and osseous abnormalities deserve particular attention even when they are not obvious on initial examination. Accelerated tooth formation or eruption, macrodontia, root hypoplasia, early loss of deciduous or permanent teeth and hypertrophy of the maxilla or mandible on the affected side have been reported [6,9]; Sun et al. showed that dental abnormalities are a frequent marker of the disease [6]. Osseous hyperplasia underlying the fatty infiltration is well demonstrated by CT, as in a Moroccan case reported by Harouna et al. (Figure 5) [7]. In our patient, these manifestations were not clinically prominent; nevertheless, a complete dental and osseous assessment remains advisable in children and adolescents in order to detect underestimated involvement and to plan orthodontic follow-up if required.

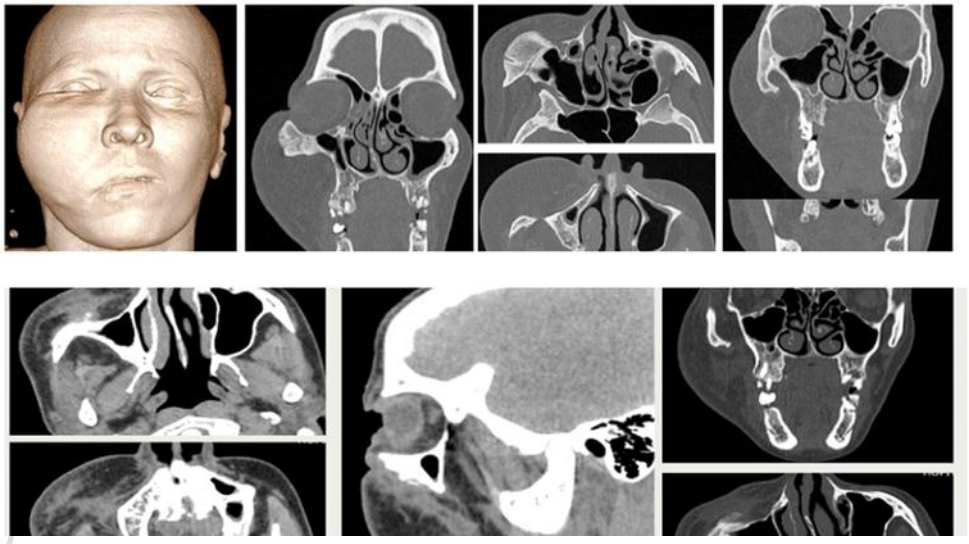

Figure 5. Moroccan case of facial infiltrating lipomatosis: three-dimensional CT reconstruction and CT slices showing soft tissue thickening of the right hemiface and hyperplasia of the underlying bones. *Reproduced from Harouna et al. [7], an open-access article distributed under the Creative Commons Attribution 4.0 International License (CC BY 4.0).*

The differential diagnosis includes simple lipoma, lipoblastomatosis, vascular or lymphatic malformations, hemifacial hyperplasia, Proteus syndrome, other lesions of the PIK3CA-related overgrowth spectrum (PROS) and, more rarely, malignant adipose tumours [9–11]. Slow progression, soft consistency, absence of pulsatility, fatty signal on MRI and absence of histological atypia point towards a benign lesion, whereas the absence of a capsule and the

infiltration of the deep planes distinguish infiltrating lipomatosis from a conventional lipoma. In our case, the lesion was neither compressible nor pulsatile, showed no vascular enhancement and infiltrated the buccal planes, making a vascular malformation unlikely.

Understanding of the pathophysiology has advanced considerably through genetic studies. Activating somatic mutations of *PIK3CA*, the gene encoding the catalytic alpha subunit of phosphatidylinositol 3-kinase (PI3K), have been identified in affected tissues [10]. These mutations activate the PI3K–AKT–mTOR pathway, which drives cell growth and tissue proliferation, and place the disease within the PROS [8,11]. This explains the variability of presentations, ranging from localised buccal involvement to extensive hemifacial forms with bone abnormalities, neurological involvement or associated malformations. No molecular analysis was performed in our patient; it could be considered in the event of recurrence, extension, a syndromic presentation or with a view to targeted therapy.

Treatment is essentially surgical, but it should not be conceived as an oncological resection. The realistic objective is to reduce volume, improve symmetry and preserve function. Complete excision is often impossible because the pathological fat infiltrates muscles, salivary glands and the deep regions of the face, with a risk of injury to the facial nerve, the parotid duct or vascular structures [4,12,13]. Depending on age, extent of disease and the patient's wishes, the options include partial reduction, liposuction, soft tissue remodelling, cheiloplasty, bone contouring and orthodontic treatment, often in several stages [6,13,14]. In our case, partial piecemeal excision through a double approach was adapted to the buccal topography and to the need to preserve vital structures; the absence of facial palsy and salivary fistula supports this cautious strategy.

The risk of recurrence remains a major element of follow-up. Recurrence or regrowth is explained by the persistence of infiltrating tissue in the deep planes and by the progression of the disease during growth [2,4,12]. Some teams therefore recommend deferring aggressive procedures until the end of growth, except in the case of significant functional, social or psychological impact [6,13]. Therapies targeting the PI3K pathway represent a promising avenue for extensive or recurrent forms, but they do not currently replace surgery in localised disease [8,11]. Our patient requires prolonged follow-up focused on the stability of the aesthetic result, perioral function, facial nerve integrity and the detection of recurrence.

The interest of this report lies in documenting an isolated buccal form of CILF in a Moroccan adolescent, with typical imaging findings and conservative management. Its limitations are the single-case design, the absence of genetic testing, the lack of a detailed CT assessment of the bony and dental structures and the short postoperative follow-up. Despite these limitations, it is a reminder that CILF should be considered in any soft, unilateral, ill-defined cheek swelling progressing since childhood, especially when MRI shows diffuse fatty infiltration without significant enhancement.

Conclusion

Congenital infiltrating lipomatosis of the cheek is a rare, benign but locally infiltrative condition responsible for progressive facial asymmetry. Our case underlines the value of MRI in confirming the fatty nature of the lesion, defining its deep extension and guiding the

operative strategy. Treatment relies on cautious, often partial surgery aimed at aesthetic improvement while preserving the facial nerve and the parotid duct. Prolonged follow-up remains essential because of the risk of recurrence and of progression with growth.

Declarations

Ethics approval and consent to participate

This work was conducted in accordance with the principles of the Declaration of Helsinki. As the patient was a minor at the time of management, both the patient and his legal representative were informed and gave their consent.

Consent for publication

Written informed consent for the publication of this case report and of the accompanying clinical images was obtained from the patient's legal representative. The images were anonymised to protect the patient's identity. A copy of the written consent is available for review by the Editor-in-Chief on request.

Availability of data and materials

All data generated or analysed during this study are included in this article. Further information is available from the corresponding author on reasonable request.

Competing interests

The authors declare that they have no competing interests.

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Authors' contributions

RB: conception of the report, clinical management, data collection and drafting of the manuscript. SK and AO: surgical management, acquisition of operative data and critical revision of the manuscript. OEG: literature review, drafting of the discussion and verification of references. ZS: supervision and critical revision of the manuscript. All authors read and approved the final version of the manuscript.

Reporting guideline

This case report was prepared in accordance with the CARE guidelines.

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